

WE CLAIM:

1. An isolated nucleic acid comprising any one of SEQ ID NOS:1-32, or a sequence complementary to any one of SEQ ID NOS:1-32.
2. An isolated nucleic acid comprising at least eight consecutive nucleotides of a nucleotide sequence of any one of SEQ ID NOS:1-32, or at least eight consecutive nucleotides of a nucleotide sequence complementary to any one of SEQ ID NOS:1-32.
3. An isolated nucleic acid comprising at least 80% nucleotide identity with a nucleic acid comprising any one of SEQ ID NOS:1-32, or at least 80% nucleotide identity with a nucleic acid comprising a sequence complementary to any one of SEQ ID NOS:1-32.
4. The isolated nucleic acid according to claim 3, wherein the nucleic acid comprises at least an 85%, 90%, 95%, or 98% nucleotide identity with the nucleic acid comprising any one of SEQ ID NOS:1-32, or at least an 85%, 90%, 95%, or 98% nucleotide identity with the nucleic acid comprising a sequence complementary to any one of SEQ ID NOS:1-32.
5. An isolated nucleic acid that hybridizes under high stringency conditions with a nucleic acid comprising any one of SEQ ID NOS:1-32, or with a nucleic acid comprising a nucleotide sequence complementary to any one of SEQ ID NOS:1-32.
6. A nucleotide probe or primer specific of ABCC12 gene, wherein the nucleotide probe or primer comprises at least 15 consecutive nucleotides of a nucleotide sequence of any one of SEQ ID NOS:1-32, or at least 15 consecutive nucleotides of a nucleotide sequence complementary to any one of SEQ ID NOS:1-32.

7. A nucleotide probe or primer specific for an ABCC12 gene, wherein the nucleotide probe or primer comprises a nucleotide sequence of any one of SEQ ID NOS:35-46, or a nucleotide sequence complementary to any one of SEQ ID NOS:35-46.

8. A method of amplifying a region of the nucleic acid according to claim 1, comprising:

- a) contacting the nucleic acid with two nucleotide primers, wherein the first nucleotide primer hybridizes at a position 5' of the region of the nucleic acid to be amplified, and the second nucleotide primer hybridizes at a position 3' of the region of the nucleic acid to be amplified, in the presence of reagents necessary for an amplification reaction;
- b) amplifying the target nucleic acid; and
- c) detecting the amplified nucleic acid region.

9. The method according to claim 8, wherein each nucleic acid primer is independently selected from the group consisting of

- a) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence of any one of SEQ ID NOS:1-32,
- b) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence complementary to any one of SEQ ID NOS:1-32,
- c) a nucleotide primer as in any one of claims 6-8,

- d) a nucleotide primer comprising a nucleotide sequence of any one of SEQ ID NOS:35-46, and
- e) a nucleotide primer comprising a nucleotide sequence complementary to any one of SEQ ID NOS:35-46.

10. A kit for amplifying the nucleic acid according to claim 1, wherein the kit comprises:

- a) two nucleotide primers whose hybridization position is located respectively 5' and 3' of the region of the nucleic acid to be amplified; and optionally,
- b) reagents necessary for an amplification reaction.

11. The kit according to claim 10, wherein each nucleotide primer is independently selected from the group consisting of

- a) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence of any one of SEQ ID NOS:1-32,
- b) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence complementary to any one of SEQ ID NOS:1-32,
- c) a nucleotide primer as in any one of claims 6-8,
- d) a nucleotide primer comprising a nucleotide sequence of any one of SEQ ID NOS:35-46, and
- e) a nucleotide primer comprising a nucleotide sequence complementary to any one of SEQ ID NOS:35-46.

12. The nucleotide probe or primer according to any one of claims 6-8, wherein the nucleotide probe or primer further comprises a marker compound.

13. A method of detecting a nucleic acid according to claim 1, wherein the method comprises:

- a) contacting the nucleic acid to be detected with a nucleotide probe selected from the group consisting of
  - i) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence of any one of SEQ ID NOS:1-32,
  - ii) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence complementary to any one of SEQ ID NOS:1-32,
  - iii) a nucleotide primer as in any one of claims 6-8,
  - iv) a nucleotide primer comprising a nucleotide sequence of any one of SEQ ID NOS:35-46, and
  - v) a nucleotide primer comprising a nucleotide sequence complementary to any one of SEQ ID NOS:35-46; and
- b) detecting a complex formed between the nucleic acid and the probe.

14. The method of claim 13, wherein the probe is immobilized on a support.

16. A kit for detecting the nucleic acid according to claim 1, wherein the kit comprises

- a) a nucleotide probe selected from the group consisting of
  - i) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence of any one of SEQ ID NOS:1-32,
  - ii) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence complementary to any one of SEQ ID NOS:1-32,
  - iii) a nucleotide primer as in any one of claims 6-8,
  - iv) a nucleotide primer comprising a nucleotide sequence of any one of SEQ ID NOS:35-46, and
  - v) a nucleotide primer comprising a nucleotide sequence complementary to any one of SEQ ID NOS:35-46; and optionally,
- b) reagents necessary for a hybridization reaction.

16. The kit according to claim 15, wherein the probe is immobilized on a support.

17. A recombinant vector comprising the nucleic acid according claim 1.

18. The vector according to claim 17, wherein the vector is an adenovirus.

19. A recombinant host cell comprising the recombinant vector according to claim 17.

20. A recombinant host cell comprising the nucleic acid according claim 1.
21. An isolated nucleic acid encoding a polypeptide comprising an amino acid sequence of any one of SEQ ID NO:33 or SEQ ID NO:34.
22. A recombinant vector comprising the nucleic acid according to claim 21.
23. A recombinant host cell comprising the nucleic acid according to claim 21.
24. A recombinant host cell comprising the recombinant vector according to claim 22.
25. An isolated polypeptide selected from the group consisting of
  - a) a polypeptide comprising an amino acid sequence of SEQ ID NO:33 or SEQ ID NO:34,
  - b) a polypeptide fragment or variant of a polypeptide comprising an amino acid sequence of SEQ ID NO:33 or SEQ ID NO:34, and
  - c) a polypeptide homologous to a polypeptide comprising an amino acid sequence of SEQ ID NO:33 or SEQ ID NO:34.
26. An antibody directed against the isolated polypeptide according to claim 25.
27. The antibody according to claim 26, wherein the antibody comprises a detectable compound.

28. A method of detecting a polypeptide, wherein the method comprises

- a) contacting the polypeptide with an antibody according to claim 26; and
- b) detecting an antigen/antibody complex formed between the polypeptide and the antibody.

29. A diagnostic kit for detecting a polypeptide, wherein the kit comprises

- a) the antibody according to claim 26; and
- b) a reagent allowing detection of an antigen/antibody complex formed between the polypeptide and the antibody.

30. A pharmaceutical composition comprising the nucleic acid according to claim 1 and a physiologically compatible excipient.

31. A pharmaceutical composition comprising the recombinant vector according to claim 17 and a physiologically compatible excipient.

32. A method of treating and/or preventing paroxysmal kinesigenic choreoathetosis in a subject in need thereof by administering the nucleic acid according to claim 1.

33. A method of treating and/or preventing paroxysmal kinesigenic choreoathetosis in a subject in need thereof by administering the recombinant vector according to claim 20.

34. A method of treating and/or preventing paroxysmal kinesigenic choreoathetosis in a subject in need thereof by administering an isolated ABCC12 polypeptide comprising the amino acid sequence of SEQ ID NO:33 or SEQ ID NO:34.

35. A pharmaceutical composition comprising a polypeptide comprising an amino acid sequence of SEQ ID NO:33 or SEQ ID NO:34, and a physiologically compatible excipient.

36. A method of identifying active ingredients for the prevention or treatment of paroxysmal kinesigenic choreoathetosis using an isolated ABCC12 polypeptide comprising an amino acid sequence of SEQ ID NO:33 or SEQ ID NO:34.

37. A method of identifying active ingredients for the prevention or treatment of paroxysmal kinesigenic choreoathetosis using a recombinant host cell expressing the ABCC12 polypeptide comprising an amino acid sequence of SEQ ID NO:33 or SEQ ID NO:34.

38. A method of screening an agonist or an antagonist of the ABCC12 polypeptide, comprising

- a) preparing a membrane vesicle comprising at least one of the ABCC12 polypeptide and a substrate comprising a detectable marker;
- b) incubating the vesicle obtained in step a) with an agonist or antagonist candidate compound;
- c) qualitatively and/or quantitatively measuring a release of the substrate comprising the detectable marker; and
- d) comparing the release of the substrate measured in step b) with a measurement of a release of a labeled substrate by a membrane vesicle that has not been previously incubated with the agonist or antagonist candidate compound.



39. A method of screening an agonist, or an antagonist of ABCC12 polypeptide, comprising
- a) incubating a cell that expresses the ABCC12 polypeptide with an anion labeled with a detectable marker;
  - b) washing the cell of step a) whereby excess labeled anion that has not penetrated into the cell is removed;
  - c) incubating the cell obtained in step b) with an agonist or antagonist candidate compound for the ABCC12 polypeptide;
  - d) measuring efflux of the labeled anion from the cell; and
  - e) comparing the efflux of the labeled anion determined in step d) with efflux of a labeled anion measured with a cell that has not been previously incubated with the agonist or antagonist candidate compound.
40. An implant comprising the recombinant host cell according to claim 23 or 24.